System components and tunable parameters

Program	Function performed
BlastParse.pl	Parse BLAST output
mark_substitution_heteroplasmy.pl	Mark ambiguous base calls
extract_SE_consensus.pl	Extract a consensus sequence from a phrap polyphred run, replacing low quality bases with '?', and polyphred rank 1 bases with 'N'.
compute_coverage.pl	Calculates overall, forward strand, and reverse strand coverage for all bases in the consensus sequence
count_hv1_deletes.pl	Count deletions in HV1 relative to the rCRS
border_index.pl	Compute start and end positions of HV1 and HV2 regions.
fix_mitotype_reporting_range.pl	Output a mtDNA profile based on input variants list and reportable range.
find_bad_traces_from_blast_report.pl	Reject reads that do not align appropriately to the rCRS
determineReadTypes.pl	Adds template name, template type, and primer type to phred output files.
seq2delta_vs	Align mtDNA profile to rCRS and report variants according to nomenclature.
calculate_coverage_mitotype.pl	Mask mtDNA consensus sequence based on required coverage and calculated coverage
count_hv1_inserts.pl	Count insertions in HV1 relative to the rCRS
generate_hv_mask_fasta_files.pl	Extract the HV1 portion and HV2 portion of the mtDNA consensus sequence, based on the computed HV1 and HV2 regions.
flip_fasta.pl	Invert an X masked sequence output by cross_match

FIG. 5

	Assessment 1	Assessment 2
Total True Positives: $N_{automated} = N_{manual}$	16	10
Total True Negatives: $A_{automated} = A_{manual}$	77,358	110,354
Total False Positives: $N_{automated} = A_{manual}$	95	807
Total False Negatives: $A_{automated} = N_{manual}$	6*	14
Total Incorrect	0	0
Sensitivity: TP/(TP+FN)	72.73%	41.67%
Specificity: TN/(TN+FP)	99.88%	99.27%
Positive Predictive Value: TP/(TP+FP)	14.41%	1.22%
Negative Predictive Value: TN/(TN+FN)	99.99%	99.99%

* Predominate base correctly called
Differences take into account the consensus of the two analysts

FIG. 6